ITPKC gene

inositol-trisphosphate 3-kinase C

Normal Function

The *ITPKC* gene provides instructions for making one version (isoform) of the inositol 1,4,5-trisphosphate 3-kinase (ITPK) enzyme. This enzyme helps add a cluster of oxygen and phosphorus atoms (a phosphate group) to a molecule called Ins(1,4,5)P3 to produce a molecule called Ins(1,3,4,5)P4. Both of these molecules are involved in regulating the amount of calcium in cells.

Several versions (isoforms) of the ITPK enzyme are produced from different genes. They play a variety of roles in processes throughout the body. The isoform produced from the *ITPKC* gene is called inositol 1,4,5-trisphosphate 3-kinase C (ITPKC). It is involved in a mechanism called the Ca(2+)/NFAT signaling pathway, which is affected by calcium levels. This pathway helps limit the activity of immune system cells called T cells. T cells identify foreign substances and defend the body against infection. Reducing the activity of T cells when appropriate prevents the overproduction of immune proteins called cytokines that lead to inflammation and which, in excess, cause tissue damage.

Health Conditions Related to Genetic Changes

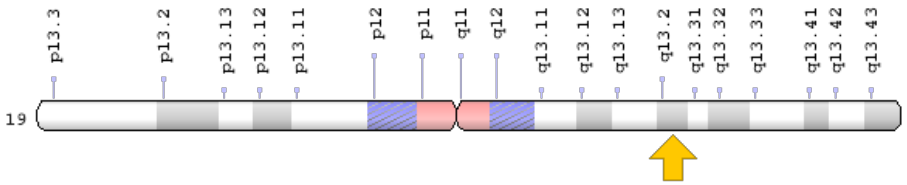
Kawasaki disease

A variation in the *ITPKC* gene has been associated with an increased risk of Kawasaki disease, a sudden and time-limited (acute) illness affecting infants and children resulting in prolonged fever, inflammation, and other signs and symptoms. The variation changes a single DNA building block (nucleotide) in a region of the gene known as intron 1. It appears to reduce the efficiency of *ITPKC* gene transcription, which is the first step in producing the ITPKC enzyme. Researchers suggest that the variation may reduce the amount of ITPKC enzyme and interfere with the body's ability to limit T cell activity, leading to inflammation that damages blood vessels and results in the signs and symptoms of Kawasaki disease.

Chromosomal Location

Cytogenetic Location: 19q13.2, which is the long (q) arm of chromosome 19 at position 13.2

Molecular Location: base pairs 40,717,103 to 40,740,860 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- inositol 1,4,5-trisphosphate 3-kinase C
- InsP 3 kinase C
- insP 3-kinase C
- IP3 3-kinase C
- IP3-3KC
- IP3K C
- IP3KC
- IP3KC_HUMAN

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ITPKC%5BTIAB%5D%29+OR+%28%28IP3-3KC%5BTIAB%5D%29+OR+%28IP3K+C%5BTIAB%5D%29+OR+%28InsP+3+kinase%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- INOSITOL 1,4,5-TRISPHOSPHATE 3-KINASE C
  http://omim.org/entry/606476

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ITPKC.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ITPKC%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=14897
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/80271
- UniProt
  http://www.uniprot.org/uniprot/Q96DU7

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